

Infertility

Case 1 (Master 5/2014)

A 32 years old male presented with 1ry infertility for 5 years. Semenogram revealed azoospermia with a decreased level of FSH, LH & testosterone on hormonal assay. On examination the patient is obese & anosmic.

- a) What is the name & trait of this syndrome?
- b) What other anomalies may be found in this syndrome?
- c) What is the role of defected gene in the pathogenesis of this syndrome?
- d) How you deal with this case?

Answer:

- a) Kallman syndrome (autosomal recessive or X-linked recessive)

Key points:

- 1ry infertility
- Azoospermia
- Decreased level of FSH, LH & testosterone
- Obese
- **Anosmic**

- b) Other anomalies:

- Testicular diameter less than 2 cm
- Cleft palate
- Cleft lip
- Color blindness
- Congenital deafness
- Cryptorchidism
- Obesity
- Osteopenia
- Gynecomastia
- Micropenis

- c) The gene responsible for the X-linked form is called (KALIG-1) (Kallman syndrome interval gene-1). Its function is to guide the migration of the GnRH neurons from their original site in the olfactory area to the hypothalamus. The syndrome is caused by failure of this migration & failure to secrete GnRH. The

same migratory defect affects the olfactory neurons in the nose with failure of the formation of the olfactory bulb.

d) Management:

- Pulsatile GnRH therapy: 5-20 µg per pulse / 120 min by spontaneous injection or by external mimipump
- Combined HCG (1000 – 3000 IU twice weekly IM) & HMG (150 IU 3 times/ week IM)
- The therapy should be continued till the sperms appear in the semen or occurrence of pregnancy in the wife.

Case 2 (Master 5/2015)

A 30 years old male, married 3 times with no children or abortions from these previous marriages. His semen analysis showed azoospermia. On examination he showed abnormal body measurements & physique.

- a) What is your diagnosis & DD?
- b) What is the cause of this condition?
- c) What is the full clinical picture?
- d) How to manage this case (investigation & tt)?

Answer:

a) Klinefelter syndrome

Key points:

- No children or abortions (primary infertility)
- Azoospermia
- **Abnormal body measurements**

DD: Other causes of azoospermia:

Pretesticular:

- Hypopituitarism
- Hyperprolactinemia
- Testosterone

Testicular:

- Klinefelter syndrome
- Cryptorchidism
- Sertoli cell only syndrome
- Infection (mumps orchitis)
- Surgery/trauma
- Radiation

Post-testicular:

- Vasectomy
- Agenesis of vas
- Obstructive azoospermia

b) Genetic disorder: presence of 2 or more X chromosomes plus at least one Y chromosome.

The commonest form 47 XXY. Other forms: 48 XXYY, 48XXXYY, 49XXXXYY, 46XY/47XXY.

c) Full clinical picture:

History:

- Puberty may occur at the normal range of age but it may not be complete.
- Main presentation is infertility; they may have also low desire & ED
- There may be mental retardation & psychiatric disturbances

Examination:

- Defective virilization, patients are tall & their legs are longer than the trunk.
- Testes are firm & small
- There may be gynecomastia

Associations:

- Low testosterone → osteoporosis & decreased muscular length
- Obesity, DM, varicose veins & leg ulcers
- High incidence of breast cancer

d) Management:

Investigations:

- Lab: semen is azoospermic or severe oligozoospermia in mosaic cases. ↑ FSH, LH, E & ↓ T
- Testicular biopsy: tubular fibrosis & hyalinization. Few tubules may show Sertoli cell only or spermatogenic foci in mosaic cases.
- Chromosomal study

Treatment:

- Androgen replacement therapy: as early as possible, life long
- Gynecomastia: mastectomy
- Infertility: in pt has spermatogenic foci, TESE followed by ICSI.

Case 3 (Master 12/2010)

A child 5 years old presented with an empty scrotum, the testis was not palpable in the inguinal canal.

- a) What is your diagnosis?
- b) Mention the types of clinical presentations?
- c) Write about the possible causes of this case?
- d) Mention the expected complications?
- e) Give an account on the investigations needed & treatment?

Answer:

a) Cryptorchidism

Key points:

- Empty scrotum

b) Types (with short notes):

- Absent testis
- Undescended testis
- Retractable testis
- Ectopic testis

c) Causes of this case:

- Absent testis: intrauterine infection, torsion or trauma
- Undescended testis: genetic, mechanical or hormonal factors
- Ectopic testis: misdirected into new ectopic site

d) Complications of this case (with short notes):

- Infertility
- Malignancy
- Torsion

e) Management:

Investigations:

- Chromosomal studies: may detect associated syndromes
- Diagnostic imaging: MRI, CT, US
- Diagnostic laparoscopy
- Diagnostic laparotomy

Treatment:

Medical:

- Intramuscular HCG: 500 IU twice weekly for 5 weeks
- Intranasal GnRH: one aerosol application of 200 µg into each nostril 3 times daily for 4 weeks.

Surgical:

Indications: failed medical ttt, associated anatomical abnormalities, ectopic testis

- Immediate orchidopexy
- Two-stages orchidopexy
- orchidectomy

Case 4 (Master)

A 9 month old infant presented by empty scrotum.

- a) What are the causes of empty scrotum?
- b) What are the possible associated anomalies of undescended testis?
- c) What are the complications?
- e) When & what is the treatment?

Answer:

a) Cryptorchidism

- Absent testis
- Undescended testis
- Retractable testis
- Ectopic testis

b) Inguinal hernia

c) Complications (with short notes):

- Infertility
- Malignancy
- Torsion

e) Treatment:

After 1 year to allow spontaneous descent & before the age of 2 years to prevent spermatogenic damage.

Medical:

- Intramuscular HCG: 250 IU twice weekly for 5 weeks
- Intranasal GnRH: one aerosol application of 200 µg into each nostril 3 times daily for 4 weeks.

Surgical:

Indications: failed medical ttt, associated anatomical abnormalities, ectopic testis

- Immediate orchidopexy
- Two-stages orchidopexy
- Orchiectomy

Case 5 (Master 5/2014)

A 6 years old boy was brought about by his mother who was annoyed by the observation that testes are sometimes not felt in the scrotum while she help him in shower taking. On examination: both testes were felt above & medial to the external ring. They were easily manipulated to normal scrotal position.

- a) What is the cause of this condition?
- b) List the other causes of empty scrotum?
- c) How to differentiate clinically between this case & other types?
- d) How you deal with this case?

Answer:

- a) Retractable testes: due to strong cremasteric reflex
- b) Absent testis, Undescended testis, Ectopic testis
- c) Examination in squatting position:

The patient is sitting with the hips flexed & abducted in order to eliminate the cremasteric reflex. Retractable testis easily pulled into the scrotum while undescended testis cannot be pulled.

The skin of the scrotum is well developed in retractile testes but not in undescended testes.

d) Treatment:

Medical:

- Intramuscular HCG: 500 IU twice weekly for 5 weeks
- Intranasal GnRH: one aerosol application of 200 µg into each nostril 3 times daily for 4 weeks.

Surgical:

Indications: failed medical ttt, associated anatomical abnormalities, ectopic testis

- Immediate orchidopexy
- Two-stages orchidopexy
- Orchiectomy

Case 6 (Diploma 11/2014)

Examination of one day old full term boy, the right testis was identified while the left one was impalpable even in inguinal canal the rest of examination was normal.

a) What is your probable clinical diagnosis?

b) What are the clinical DD?

c) What are the complications?

d) How can you manage this case?

Answer:

a) Cryptorchidism

b)

- Absent testis
- Undescended testis

c) Complications (with short notes):

- Infertility
- Malignancy
- Torsion

d) see before

Case 7 :

A 17 years old male patient came to out-patient clinic after vigorous exercise, with nausea, vomiting, severe unilateral scrotal pain& lower abdominal pain. The scrotal skin was severely tender, edematous & erythematous. The spermatic cord was thickened.

- a) Write about the DD?
- b) What is your diagnosis?
- c) How will you confirm your diagnosis?

Answer:

a) Acute scrotal swellings:

- Traumatic: torsion, hematocele , strangulated hernia
- Inflammatory: epididymo-orchitis
- Neoplastic: hemorrhage inside the tumor

b) Testicular torsion

c) Confirm the diagnosis:

- Diagnostic imaging: radioisotopes scanning & color doppler
- Surgical exploration: the single most important diagnostic procedure

Case 8 (Master 5/2015)

A 25 years old single male had semen analysis before going to be recruited. It showed severe oligoteratozoospermia. On examination no physical abnormalities were noticed apart from angiokeratoma of the scrotum.

- a) What is your diagnosis & DD?
- b) What is the cause of this condition?
- c) What is the full clinical picture?
- e) How to manage this case (investigation & tt)?

Answer:

- a) Subclinical varicocele

Key points:

- Severe oligoteratozoospermia (stress pattern)
- Angiokeratoma of the scrotum: thought to be associated with varicocele

DD:

- Fever
- Infections
- Radiation
- Spermicidal agents

b) Theories:

- Heat theory
- Metabolites theory
- Epididymal theory
- Intratesticular obstruction theory
- Immunological theory
- Endocrine theory

c) Full clinical picture:

1. Patients present with infertility &/or dull aching pain after prolonged standing that is relieved in the supine position.
2. Palpation of the spermatic cord reveals the varicocele like a bag of worms while pt is standing.
3. An impulse may be felt during Valsalva maneuver.
4. Examination in supine position, if it does not diminish mechanical obstruction may be present.
5. Clinical grading:

- Grade 1: palpable during valsalva maneuver only
- Grade 2: palpable without valsalva maneuver
- Grade 3: palpable & visible through scrotal skin
- Subclinical varicocele: not felt clinically but can be demonstrated by imaging

d) Investigations (with sort notes):

Diagnostic imaging:

- Color Flow Doppler Ultrasonography
- Venography
- Others: Doppler, radionuclide imaging, scrotal thermography

Laboratory:

1- Semen evaluation:

- Semen analysis: stress pattern; increased immature sperms, decreased sperm motility & concentration.
- Semen culture: high incidence of male accessory gland infection.
- Sperm function tests: abnormal zona-free hamster ova penetration test, abnormal hypo-osmotic swelling test

2-Endocrinal evaluation:

- Basal hormonal testing: \uparrow FSH, \downarrow T
- Dynamic hormonal testing: GnRH stimulation test, HCG stimulation test

Treatment:

varicocelectomy operation (with short notes)

Case 9 (Diploma 4/2016)

Male patient 38 years old complaining of scrotal pain with 2ry infertility for 5 years. Examination revealed bilateral palpable veins through the scrotal skin.

- a) What is your diagnosis?
- b) What are the investigations?
- c) How can you treat this patient?

Answer:

- a) Varicocele

Key points:

- Bilateral palpable veins through the scrotal skin
- Pain with 2ry infertility

- b) see before

- c) see before

Case 10 (Diploma 5/2014)

A 28 years old male patient suffering from 1ry infertility. His semenogram shows oligoathenozoospermia. On examination, there was a left sided varicocele.

- a) What is the prevalence of varicocele in fertile & non-fertile population?
- b) Mention the theories explaining the pathophysiology of varicocele?
- c) Why varicocele is more common on the left side?
- d) What are the indications of varicocelectomy?

Answer:

a)

b) Theories (with short notes)

- Heat theory
- Metabolites theory
- Epididymal theory
- Intratesticular obstruction theory
- Immunological theory
- Endocrine theory

c) Varicocele is more common on the left side:

- Left testicular vein is longer than the right testicular vein.
- Left testicular veins have less valves.
- Left testicular vein drains at right angle into the left renal vein while the right testicular vein drains obliquely into IVC
- Left renal vein may be compressed from outside (nutcracker phenomenon)

d) Indications of varicocelectomy:

- Infertile patients with clinical varicocele associated with subnormal testicular size & semen parameters.
- Adolescent boys with clinical varicocele associated with more than 20% reduction in the testicular volume &/or failure of the testicular growth on repeated follow up from year to year.
- Patients with clinical varicocele associated with debilitating testicular pain.

Case 11 (Master 12/2013)

A 34 years old male patient complaining of 1ry infertility. His semen parameters revealed asthenoteratozoospermia. On examination, there was a left sided varicocele.

- a) What is the prevalence of varicocele in fertile & non-fertile population?
- b) Mention the theories explaining the pathophysiology of varicocele ?
- c) What varicocele is more common on the left side?
- d) What are the indications of varicocelectomy?

Answer:

See before

Case 12 (Master 5/2015)

A 27 years old married male looked for advice for his fertility problem. . His semen analysis showed azoospermia. His hormonal pattern was normal as regards FSH, LH, prolactin, testosterone, estrogen. On examination his epididymis was full, no varicocele was detected.

- a) What is your diagnosis & DD?
- b) What is the cause of this condition?
- c) What is the full clinical picture?
- e) How to manage this case (investigation & tt)?

Answer:

- a) Obstructive infertility (epididymal obstruction)

Key points:

- Epididymis was full
- Azoospermia
- Hormonal pattern was normal

DD: Other causes of azoospermia: see before

- b) Epididymal obstruction

c)

- d) Management:

- Quantitative analysis of testicular biopsy.

- If the patient has a testicular biopsy with normal spermatogenesis & palpable vas with azospermic ejaculate → obstruction & is indicated for scrotal exploration & possible epididymovasostomy.

- Testicular &/or epididymal sperm retrieval followed by ICSI is indicated if surgical correction is failed or impossible.

Case 13 (Diploma 11/2015)

A 37 years old male presented with 2ry infertility for 6 years. Clinical examination showed bilateral epididymal tail nodules & semen analysis azoospermic with normal volume & leukocytospermia. There was history of recurrent urinary tract infections.

- a) What is your diagnosis?
- b) Mention his hormonal profile?
- c) How can you treat this patient?

Answer:

- a) Obstructive infertility (epididymal obstruction)

Key points:

- 2ry infertility
- Bilateral epididymal tail nodules
- Azoospermic
- Leukocytospermia (infection)
- Recurrent urinary tract infections

- b) Hormonal profile: Normal

c) Treatment:

- Treatment of genital infection
- Epididymovasostomy.
- Testicular &/or epididymal sperm retrieval followed by ICSI is indicated if surgical correction is failed or impossible.

Case 14 (Diploma 11/2012)

A 35 years old man presented with 1ry infertility for 3 years, his semen profile was azoospermic with normal volume, testosterone, LH, FSH, also testicular size was normal.

a) What is your diagnosis?

b) How can you manage this case?

Answer:

a) Obstructive infertility

Key points:

- Azoospermic
- Normal volume, testosterone, LH, FSH, also testicular size was normal.

b) Quantitative analysis of testicular biopsy.

If the patient has a testicular biopsy with normal spermatogenesis & palpable vas with azoospermic ejaculate → obstruction & is indicated for scrotal exploration & possible epididymovasostomy.

Testicular &/or epididymal sperm retrieval followed by ICSI is indicated if surgical correction is failed or impossible.

Case 15 (Master 5/2015)

A 35 years old married male who works in the field of manufacturing cellular phones had 2 children of 9 years of marriage. For the last 5 years he was trying to get his wife pregnant but failed to do so.

- a) What is your diagnosis & DD?
- b) What is the cause of this condition?
- c) What is the full clinical picture?
- d) How to manage this case (investigation & treatment)?

Answer:

- a) Secondary infertility due to irradiation

Key points:

- Had 2 children (secondary)
- Field of manufacturing cellular phones (gonadotoxins)

DD: Other causes of gonadotoxins:

- b) Irradiation in the field of manufacturing cellular phones is gonadotoxin.
- c)
- d)

Case 16 (Master 4/2012)

A 33 years old male complaining from primary infertility 3 years ago. Semen parameters revealed low semen volume (0.5 ml), low PH & azoospermia..

a) What is your diagnosis?

b) What is your management?

Answer:

a) Congenital absent vas

Key points

- Primary infertility
- Low semen volume
- **Low PH & azoospermia**

b) Management

- Genetic counselling is essential before ICSI
- Testicular &/or epididymal sperm retrieval followed by ICSI